Case Report

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Delayed Diagnosis of Hemophilia B Presenting with Hematemesis

Abstract

A 36-year-old nonalcoholic gentleman presented with history of moderate-amount hematemesis. He received multiple blood transfusions. Upper gastrointestinal endoscopy was done which showed no evidence of varices, but a tiny bleeding ulcer was seen in lower end of oesophagus; sclerosing agent was injected at the base of ulcer. All the blood investigations were normal. A diagnosis of Dieulafoy’s lesion was made. After two months, he developed hemorrhaxis after sustaining injury in right knee. Again investigations were done and coagulation profile revealed raised activated partial thromboplastin time and normal prothrombin time. Factor VIII and IX assay were sent in view of raised aPTT. Factor VIII and IX was 120% and 4% of normal respectively. Von Willebrand factor levels were within normal limits. Revised diagnosis of moderate hemophilia B was made.

Keywords: Hemophilia, Hematemesis.

Introduction

Dieulafoy’s lesion is a rare cause of upper gastrointestinal (GI) bleeding but can cause potentially life-threatening hemorrhage, especially when associated with a coagulation disorder like hemophilia. Here we present a case that manifested with acute onset of non-variceal upper gastrointestinal bleeding at 36 years of age.

Case History

A 36-year-old engineer presented to the emergency room with history of two episodes of vomiting of altered blood, moderate in amount since last half-an-hour. He was on fast due to the festival of Ramzan. He again vomited approximately 150 mL of coffee-colored vomitus in the emergency. There was no past history of similar episode, NSAIDS use, bleeding diathesis, jaundice, anorexia, weight loss, pain, or burning sensation in epigastrium, black stools, easy bruisability, or bleeding gums. He was nonalcoholic. A detailed family history, including the first degree relatives, was negative for bleeding diathesis.

On examination, vitals were stable and general physical examination did not reveal any significant abnormality; there were no signs of chronic liver failure; systemic examination was also within normal limits. Nasogastric tube was inserted and treatment was initiated on the lines of upper gastrointestinal bleed. Intravenous colloids were given, injection Pantoprazole was given as 80 mg bolus intravenous and maintenance of 8 mg/h; injection Octreotide was given as 100 mcg IV bolus stat and at maintenance dose of 50 mcg/hr. His vitals were stable, though his gastric output was around 450 mL containing fresh blood and clots. In view of ongoing hematemesis, two units of whole blood were transfused. He was airlifted via helicopter to a tertiary care hospital where further investigations were done. Total leukocyte count was 11.000/mm³, platelet count 2.7 lac/mm³, hemoglobin 12.5 g/dL. Serum bilirubin 0.8 mg/dL (direct 0.6, indirect 0.2), SGOT 35 IU/L, SGPT 32 IU/L, blood glucose 98 mg/dL, serum urea 23 mg/dL, creatinine 0.8 mg/dL. Prothrombin time (test) 13 sec (control) 14 sec, activated partial thromboplastin time (test) 35 sec (control) 34 sec; clotting time 6.8 sec, bleeding time 2.5 sec. Ultrasonography for abdomen and Doppler for spleno-
portal axis did not show any significant abnormality. Upper GI endoscopy was done which showed a tiny bleeding ulcer in the lower part of esophagus (Fig. 1). There was no evidence of any esophageal or gastric varices. Sclerosing agent was injected at the base of ulcer (Fig. 2). Final diagnosis was made as upper gastrointestinal bleed was likely due to a Dieulafoy’s lesion.

Patient was asymptomatic for the next two months when he sustained a knee injury while playing badminton. The left knee was totally swollen, tender, and hot; swelling at lower part of the thigh was also present. Hemarthrosis with thigh hematoma was suspected. We again did coagulation profile which revealed prothrombin time (test) 14 sec (control) 13 sec; activated partial thromboplastin time (aPTT) (test) 62 sec (control) 34 sec; Factor VIII and IX assay were sent in view of raised aPTT. Factor VIII and IX assay was 120% and 4% of normal respectively. Von Willebrand factor levels were within normal limits. Revised diagnosis of moderate hemophilia B was made. Magnetic resonance imaging of knee revealed anterior cruciate ligament tear with hemarthrosis and hematoma in thigh. Patient was managed conservatively and discharged.

**Discussion**

Upper gastrointestinal bleeding is a common medical condition resulting in substantial morbidity and mortality. There are many causes of upper GI bleed, Dieulafoy’s lesion being one of them. It leads to less than 5% of all gastrointestinal bleeds in adults. Dieulafoy’s lesion is characterized by a large tortuous arteriole in the stomach wall that protrudes through a tiny mucosal defect and bleeds when eroded. Most of these lesions are located in the stomach and lower end of esophagus but may be found in other parts of the gastrointestinal tract, including the small bowel. Dieulafoy’s lesions are more common in males, occur at a median age of 54 years, and are not related to alcohol or non-steroidal anti-inflammatory drug use. These bleeding episodes can present as recurrent hematemesis associated with melena. These lesions can easily be missed during endoscopy due to the intermittent nature of bleeding. Approximately 49% of these lesions are identified during an initial endoscopic examination; however, 33% of lesions require more than one endoscopy for identification. The remaining cases are identified via angiography during active bleeding or via laparotomy. An endoscopic ultrasound may also confirm the diagnosis by revealing a tortuous submucosal vessel near the mucosal defect. Therapeutic endoscopy is the initial treatment of choice. Different endoscopic modalities have been used, including bipolar electrocoagulation, injection sclerotherapy, heater probe, laser photocoagulation, epinephrine injection, hemoclipping, and banding. Angiography is usually reserved for patients who are not amenable to endoscopic therapy and who are poor surgical candidates. Duodenal and proximal jejunal lesions can be managed by surgical exploration via intraoperative endoscopy.

Hemophilia is a group of inherited blood disorders in which blood does not clot properly. Hemophilia B, or Christmas disease, is an inherited, X-linked, recessive disorder that results in deficiency of functional plasma coagulation factor IX. It may appear as severe, moderate or mild hemorrhagic disease. In the severe form, it is characterized by multiple bleeding episodes in joints and other tissues leading to chronic crippling hemarthropathy unless treated early or prophylactically with factor IX concentrates. People with mild disease may not have any symptoms until some major surgery or trauma occurs. Laboratory studies for suspected
hemophilia B include a complete blood cell count, coagulation studies, and a factor IX (FIX) assay. Bleeding time and prothrombin time (which assesses the extrinsic coagulation pathway) are normal. Usually, the aPTT is prolonged; however, a normal aPTT does not exclude mild or even moderate hemophilia because of the relative insensitivity of the test. The aPTT is significantly prolonged in severe hemophilia. Reference range for Factor IX is 70-120% of normal values. In the mild disease, it is over 5% of normal value; in moderate disease it is 1-5% and in severe disease below 1% of normal values. In our case, the values of Factor IX were 4% making the disease to fall in moderate category.

This case was interesting because there was no history of bleeding manifestations, joint swellings, rashes, etc., in the past, nor was there any positive family history. Dieulafoy’s lesion is usually associated with minor bleeding episodes but here in this case, as the patient was hemophilic, the amount of bleeding was quite high. Also hemophilia was only suspected when hemorrhaxis occurred. Initial report of aPTT was normal which could have been erroneous due to history of blood transfusion a day prior and also can be there in mild variants of the disease.

**Conclusion**

Dieulafoy’s lesion is a rare but an important cause of gastrointestinal bleeding. They can easily be missed during endoscopy due to the intermittent nature of bleeding. It can be fatal if not treated early. Mild or moderate hemophilia can remain silent in childhood and can sometimes present for the first time with bleeding manifestations in adult life. Complete coagulation profile including PT, aPTT, bleeding and clotting time should always be performed to rule out bleeding disorders even in the presence of a structural lesion as the prognosis can be very different when a bleeding disorder is coexisting.

**Conflict of Interest:** None

**References**


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